Congenital Cataract- Stepwise approach to determining etiology

Preform History and Physical Examination to Determine if one of the following present

- Multiple Congenital Anomalies
- Isolated Ocular Abnormalities
- One organ system abnormality
- No other abnormalities

Congenital Cataracts and Multiple Congenital Anomalies present

Do Chromosome Analysis to screen for the following:

- Trisomy 18
- Trisomy 21
- Turner Syndrome
- Edward Syndrome
- Partial Trisomy 10q
- Translocations: 3:4, 2:14, 2:16
- Cri du Chat syndrome (5q-)

If Chromosome Analysis Normal consider:

- Hallerman-Strieff syndrome: dental anomalies
- Schprintzen syndrome: cardiac anomalies
- Cerebral-oculo-facial syndrome: microcephaly
- Cockayne syndrome: microcephaly
- Fetal rubella syndrome: microcephaly
- Marinesco-Sjögren syndrome: microcephaly
- Mowat-Wilson syndrome
- Rubenstein-Taybi syndrome: microcephaly
- Smith-Lemli-Opitz syndrome: microcephaly
- Walker-Warburg syndrome: hydrocephalus
- Zellweger syndrome: long-chain fatty acids
- Lowe Syndrome***

Congential Cataracts and Isolated Ocular Abnormalities

- Aniridia
- PHPV
- Uveitis

- Retinoblastoma

Congenital Cataracts and one organ system abnormality

Short stature or limb abnormalities

Do skeletal survey to determine:

- Short limbed dwarfism
  - Camarati-Englemann syndrome
  - Chondrodysplasia punctata
  - Hypochondrodysplasia
  - Kniest syndrome
- Bony changes
  - Fetal warfarin exposure
  - Mannosidosis
  - Marshall-Stickler syndrome
  - Schwartz-Jampel syndrome
- Limb anomalities
  - Fetal Varicella infection
  - Klippel-Trenaunay-Weber syndrome
  - Proteus syndrome
  - Roberts syndrome

Muscle

Myotonic dystrophy: Muscle wasting

Aniridia and Wilms Tumor

Hearing disorder

- Refsum disease
- Alport syndrome

Dental abnormalities

May not be noted until teeth erupt

- Canine radicomegaly
- Nance-Horan syndrome (X-linked)

- Oculo-dental digital syndrome

Skin abnormalities

- Focal dermal hypoplasia
- Gorlin syndrome
- Incontinentia Pigmenti
- Rothmund-Thompson syndrome
- Fabry disease

Nail dystrophy

- Clouston syndrome (hidrotic ectodermal dysplasia) OMIM #129500
    - normal sweat and sebaceous gland function
    - total alopecia
    - severe dystrophy of the nails
    - hyperpigmentation of the skin especially over the joints
    - normal teeth
    - strabismus
    - mental deficiency
    - clubbing of the fingers
    - palmar hyperkeratosis
    - extensive kindred of French extraction that migrated to Canada, Scotland, and northern United States
- Nail-Patella syndrome OMIM #161200
    - dysplasia of the nails
    - absent or hypoplastic patellae (60-90%)
    - abnormality of the elbows interfering with pronation and supination (60-90%)
    - nephropathy
    - hearing loss
    - keratoconus
    - glaucoma, microcornea, microphakia
- Pachyonychia congenita syndrome OMIM #167200
    - onychogryposis
    - hyperkeratosis of the palms, soles, knees and elbows
    - tiny cutaneous horns in many areas
    - eukoplakia of the oral mucous membranes
    - Hyperhidrosis of the hands and feet

Congenital Cataracts without any other abnormality

Do a screen for the following:

- CBC: elevated white count

- Blood Glucose or HbA1C: hyperglycemia
- RBC Galactokinase serum level: galactosemia
- TORCH titer
    - IgG (maternal may last 6 months) and consider IgM if signs of inflammation
    - Toxoplasma- IgG +/- IgM
    - Syphilis- total antibodies with reflex confirmation
    - Rubella- IgG +/- IgM
    - Cytomegalovirus IgG +/- IgM
    - Herpes- HSV 1&2 specific IgG +/- IgM

If screen above is normal: Early onset Cataract genetic testing program by Travere Therapeutics (18 months to 35 years old with infection causes ruled out): LINK

Consider Genetics referral if syndrome above suspected

cataract